

Detection of expanded 12-mer repeat in  
NHLRC1 gene causing Lafora epilepsy in  
Beagles and Miniature Wirehaired  
Dachshunds

**Sample**

Sample: 18-06030  
Name: Jack Sparrow del Fiore Selvatico  
Breed: Beagle  
Tattoo number: N/A  
Microchip: 380 260 002 854 012  
Reg. number: LO17100227  
Date of birth: 15.04.2017  
Sex: male  
Date received: 14.03.2018  
Sample type: buccal swab

**Customer**

Francesco Mennone

**Result: N<sub>2</sub>/N<sub>3</sub>**

**Result codes:**

- N<sub>2</sub>/N<sub>2</sub>, N<sub>3</sub>/N<sub>3</sub>, N<sub>2</sub>/N<sub>3</sub> = negative genotype, dog carrying two or three 12-mers.
- N<sub>2</sub>/P, N<sub>3</sub>/P = carrier of Lafora epilepsy.
- P/P = dog affected by Lafora epilepsy.

**Explanation**

Presence or absence of expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy in Beagles and Miniature Wirehaired Dachshunds was tested. Generally, the clinical signs appear at 5-6 years of age or later. Epileptic seizures include mainly sudden involuntary muscle jerking. Over time seizures are accompanied by other neurological symptoms such as ataxia, twinkling, blindness or dementia. This form of epilepsy is incurable and fatal.

Expanded 12-mer repeat in NHLRC1 gene causing Lafora epilepsy is inherited as an autosomal recessive trait. That means the disease affects dogs with P/P genotype only. Carriers are healthy without symptoms of epilepsy. In offspring of two carriers following genotype distribution can be expected: 25 % N/N, 25 % P/P and 50 % N/P.

Method: SOP187-Lafora, ASA-PCR of DNA modified template

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Responsible person: Mgr. Martina Šafrová, Laboratory Manager



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